Case Report

Trichorhinophalangeal syndrome type 1: A case report with literature review

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Abstract

Trichorhinophalangeal syndrome is a very rare genetic disorder, where damage and mutation to the number 8 chromosome affects sufferers in numerous ways. The syndrome has three types, all characterized by abnormally short stature, sparse hair, short deformed fingers with cone-shaped epiphyses visible in radiographs. Type I is the most common. Type II is characterized by the development of multiple bony exostoses and frequently, mental disability. Type III is a more severe form of type I and is associated with short stature. This report presents a 28-year-old man who had the characteristic features of type I with the presence of multiple erupted supernumerary teeth with normal mentation and karyotyping with high resolution G banding displayed normal chromosomal complements.

Key words: Cone-shaped epiphyses, chromosome 8q 24.12, Herthoge's sign, supernumerary teeth, trichorhinophalangeal syndrome type 1

INTRODUCTION

Giedion[1] in 1966 coined the term trichorhinophalangeal syndrome type I (TRPS I) or Giedion syndrome which is an extremely rare inherited multisystem disorder characterized by thin sparse scalp hair, distinctive facial features such as laterally sparse eyebrows, bulbous tip of the nose, long flat philtrum, thin upper vermilion border and protruding ears, dental anomalies such as multiple supernumerary teeth[1] and skeletal abnormalities include cone-shaped epiphyses at the phalanges, hip malformations, and short stature.[2] TRPS I is usually inherited as a dominant trait;[3] however, it has been suggested that TRPS I may be an autosomal recessive trait[4] and is caused by deletion of the chromosome band 8q 24.12.[5]

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Trichorhinophalangeal syndrome type II or Langer–Giedion syndrome differs from type I by the presence of multiple bony or cartilagenous exostoses, and all patients show *de novo* deletion of chromosome 8q 24.12. Many patients are mildly to moderatly mentally retatarded.[6]

Trichorhinophalangeal syndrome Type III or Sugio–Kajii syndrome, a severe form of TRPS I, with severe short stature and brachydactyly.[7] It differs from TRPS II by the presence of normal intelligence and absence of exostoses and from TRPS I by the presence of severe shortness of all phalanges, metacarpals, and stature.[8]

CASE REPORT

A 28-year-old gentleman came to the Department of Oral and Maxillofacial Surgery as an outpatient with a chief complaint of pain in the upper back teeth since a week and painless swelling of his fingers since 13 years. His past history was otherwise unremarkable. His parents were not from related families and appeared normal. On examination, grossly decayed tooth due to caries was present and the extraction of the same was done under local anesthesia. Careful intra-

oral examination revealed the presence of multiple erupted supernumerary teeth (hyperdontia) in relation to upper and lower jaws. Right upper quadrant revealed the presence of fully erupted four pre-molar and four molar teeth [Figure 1a]. Left upper quadrant presented three pre-molar teeth [Figure 1b], and left lower quadrant showed the presence of completely erupted five pre-molar teeth [Figure 1c]. The hard palate was mildly high arched.

Extra oral examination revealed severe alopecia to complete baldness of the scalp. Sparse lateral eyebrows, pear-shaped nose with bulbous tip, long flat philtrum (Herthoge's sign), and thin vermilion border of upper lip, protruding ears [Figure 2a–c] and some skeletal abnormalities was present. Skeletal abnormalities included short deformed fingers and toes (clinobrachydactyly). Radial deviation of the second and third fingers and distal curvature of little fingers in both the hands with lateral bulging of the proximal interphalangeal joints was observed [Figure 3a] with bilateral racket thumb nails [Figure 2d]. His big toe was shorter than the second toe [Figure 3b]. He was short stature measuring 149 cm in height.

Radiological investigations was carried out, orthopantamography revealed three impacted teeth in relation to right lower jaw with deformed mandible [Figure 1d]. AP view of the hand demonstrated prominent cone-shaped and widened epiphyses of the second and third middle phalanges of both hands [Figure 4a]. AP view of the leg showed prominent brachydactyly of big toes [Figure 4b]. However X-ray of the pelvis declared no abnormal changes in the femoral head or neck [Figure 4c]. His mental ability was not impaired. Chromosomal analysis of the patient revealed normal karyotyping (46, XY) with no evidence of deletions and translocations [Figure 5].

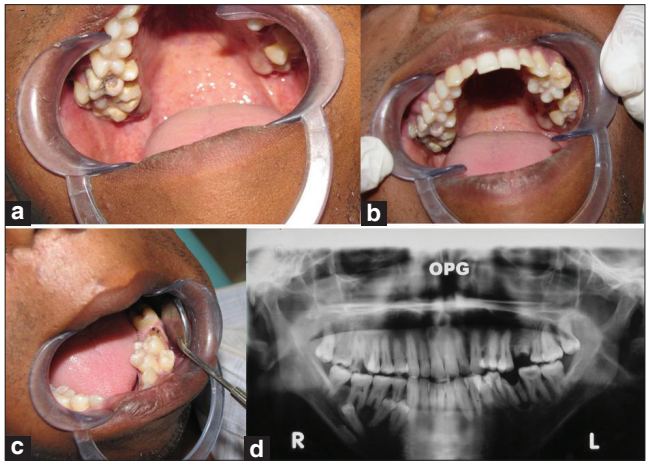

Figure 1: (a) Right upper quadrant showing the presence of fully erupted four pre-molar and four molar teeth. (b) Left upper quadrant presenting three pre-molar teeth. (c) Left lower quadrant showing the presence of completely erupted five pre-molar teeth. (d) Orthopantamography revealing three impacted teeth in relation to right lower jaw with deformed mandible

DISCUSSION

TRPS I is usually inherited as a dominant trait;[3] however, it has been suggested that TRPS I may be an autosomal recessive trait.[3,4] In our case neither of the inheritance occurred, instead the cause was a random occurrence as a result of sporadic genetic mutation with no family history of the disease. In addition, our finding of normal chromosomes agrees with the majority of the literature on TRPS I. Because 8q24.12 is a very narrow dark band, chromosomal analysis is normal in the majority of affected individuals.[9] However, Yamamoto *et al.*[10] reported a case of microdeletion of the long arm of chromosome 8q in TRPS I with severe mental retardation, but lacked multiple exostoses. The findings were interpreted to support the suggestion that TRPS II is due to deletion of 8q24.11-q24.13 and TRPS I patients have a deletion of band 8q24.12. The band 8q24.13 is involved in the development of

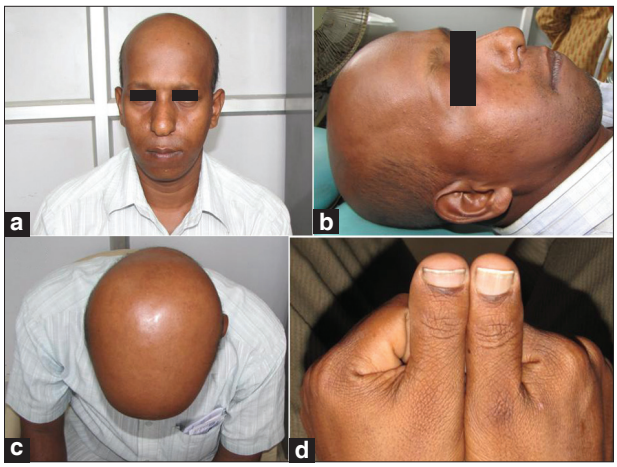

Figure 2: (a) Frontal profile showing alopecia of the scalp, pear-shaped nose with bulbous tip, long philtrum, lateral sparse eyebrow, protruding ears. (b) Lateral profile showing sparse hairs in the temporoparietal region, pear-shaped nose with bulbous tip, long philtrum, lateral sparse eyebrows. (c) Alopecia of the scalp. (d) Bilateral racket thumb nails

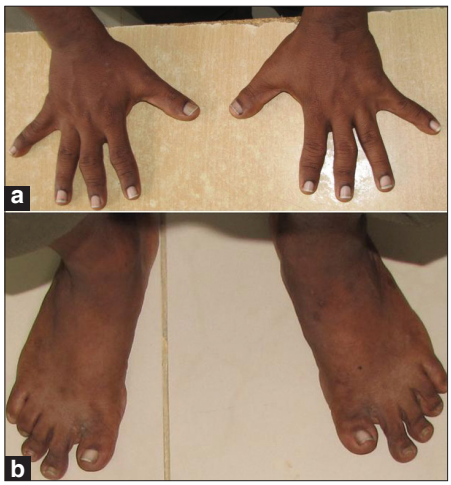

Figure 3: (a) Radial deviation of the second and third fingers and distal curvature of little fingers in both the hands with lateral bulging of the proximal interphalangeal joints. (b) Brachydactyly of the big toes

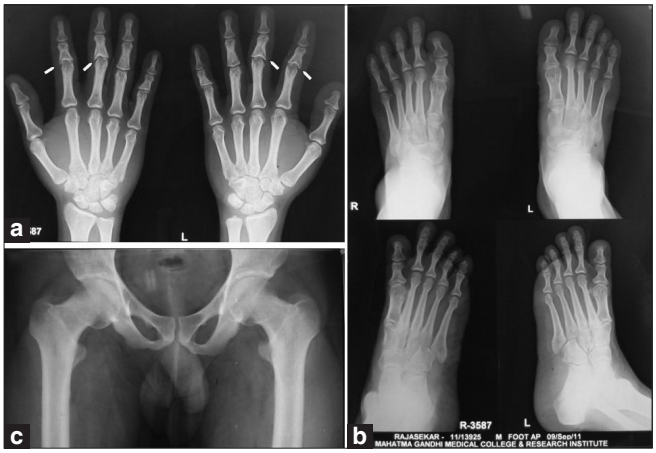

Figure 4: (a) AP view of the hand demonstrating clinobrachydactyly. White arrows indicate cone-shaped epiphyses of the middle phalanges. (b) AP view of the leg demonstrating clinobrachydactyly of big toe. (c) No evidence of Legg–Calvé–Perthes disease in the femoral head or neck

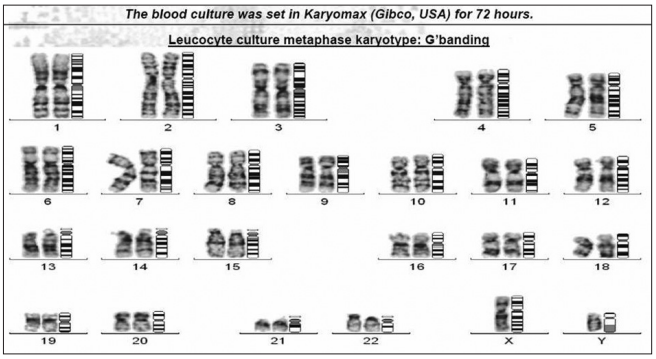

Figure 5: Karyotyping report demonstrating normal chromosomal analysis (46, XY)

exostoses and mental retardation seems to be correlated with the size of the interstitial 8q deletion. Fryns *et al.*[11] reported a case of interstitial deletion of 8q24.12 in TRPS I who had normal intelligence with no evidence of bony exostoses. Yet in another patient, Yamamoto *et al.*[12] found no evidence of deletion in a study of the chromosomes at the 850-bands stage in a patient with typical familial TRPS I which correlates with our case.

The estimate of the prevalence of TRPS I and type III in Europe was surveyed and found that more than 100 cases were published until May 2011.[13] The most typical radiographic findings are cone-shaped epiphyses, predominantly at the middle phalanges which coincide with the study conducted by Peltola *et al.*[14] who stated that all the patients described were at work until at least midlife with a normal life expectancy. Flattening of the capital femoral epiphyses, partial syndactyly, scoliosis, kyphosis, winged scapula, thoracic deformity, and mental deficiency may sometimes accompany the main features. Progressive osteoarticular changes and degenerative hip disease may necessitate orthopedic care.[9]

In our report, the patient presented sparse scanty lateral eyebrows with alopecia.[1,14] Changes in the nails were described as thin and fragile nails. Additional changes included shortening and broadening of nails giving rise to the appearance of racket nails[15] which correlates to our case. The most interesting intra-oral finding in the presented case was three impacted and seven completely erupted supernumerary teeth which pose an important clinical significance for the dentists. To our knowledge, our patient was the first one who has been reported with three impacted and seven erupted supernumerary teeth. Orthopedic considerations should be considered as first line of treatment if this type of syndrome presents with Legg–Calvé–Perthes disease.

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